Overview of Ichthyosis

Ichthyosis is a group of skin disorders that lead to dry, itchy skin that appears scaly, rough, and red. The symptoms can range from mild to severe. Ichthyosis can affect only the skin, but some forms of the disease can affect internal organs as well.

Most people inherit ichthyosis from their parents through a mutated (changed) gene. However, some people develop a form of acquired (nongenetic) ichthyosis from another medical disorder or certain medications. While there is currently no cure for ichthyosis, research is ongoing and treatments are available to help manage the symptoms.

The outlook for people with ichthyosis varies depending on the type of the disease and how severe it is. Most people with ichthyosis need treatment for life to help make the disease more manageable.

Who Gets Ichthyosis?

Anyone can get ichthyosis. The disease is usually passed down from your parents; however, some people can be the first in a family to develop ichthyosis due to a new gene mutation. Other people develop an acquired (nongenetic) form of ichthyosis, which results from another medical condition or a side effect of a medication.

Types of Ichthyosis

There are more than 20 features of ichthyosis, including those that occur as part of another syndrome or condition. Doctors may determine the type of ichthyosis by identifying the:

- Gene mutation.
- Inheritance pattern through analyzing family trees.
- Symptoms, including their severity and which organs they affect.
- Age when symptoms first appeared.

Some types of the disease, which are inherited and are not part of a syndrome, include the following:

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Ichthyosis vulgaris is the most common type. It is usually mild and appears in the first year of life with dry, flaky skin.

- Harlequin ichthyosis is usually seen at birth and causes thick scaly plates of skin that cover the entire body. This form of the disorder can affect the shape of facial features and may limit joint movement.
- Epidermolytic ichthyosis is present at birth. Most infants are born with fragile skin and blisters covering their body. Over time, the blisters disappear, and scaling of the skin develops. This can have a ridged appearance over areas of the body that bend.
- Lamellar ichthyosis is present at birth. The infant is born with a tight clear covering the entire body, called a collodion membrane. Over several weeks, the membrane peels away, and large, dark, plate-like scales develop over most of the body.
- Congenital ichthyosiform erythroderma is present at birth. Infants also often present with a collodion membrane.
- X-linked ichthyosis usually develops in males and begins at about 3 to 6 months of life. Scaling is usually present on the neck, lower face, trunk, and legs, and symptoms can worsen over time.
- Erythrokeratodermia variabilis usually develops a few months after birth and progresses during childhood. The skin can develop rough, thick or reddened areas of skin, usually on the face, buttocks, or limbs. The affected areas can spread on the skin over time.
- Progressive symmetric erythrokeratoderma usually appears in childhood with dry, red, scaly skin primarily on the limbs, buttocks, face, ankles, and wrists.

Symptoms of Ichthyosis

The symptoms of ichthyosis can range from mild to severe. The most common symptoms include:

- Dry skin.
- Itching.
- Redness of the skin.
- Cracking of the skin.
- Scales on the skin that are white, gray, or brown and have the following appearance:
  - Small and flaky.
  - Large, dark, plate-like scales.
  - Hard, armor-like scales.

Depending on the type of ichthyosis, other symptoms may include:

- Blisters that can break, leading to wounds.

- Hair loss or fragile hair.
- Dry eyes and difficulty closing eyelids. Inability to perspire (sweat) because skin scales clog the sweat glands.
- Difficulty hearing.
- Thickening of the skin on the palms of the hands and soles of the feet.
- Tightening of the skin.
- Difficulty flexing some joints.
- Open wounds from scratching itchy skin.

Cause of Ichthyosis

Gene mutations (changes) cause all of the inherited types of ichthyosis. Many gene mutations have been identified and the inheritance pattern depends on the type of ichthyosis. People continually grow new skin and shed old skin throughout their lives. For people with ichthyosis, the mutated genes change the normal skin growth and shedding cycle, causing skin cells to do one of the following:

- Grow faster than they are shed.
- Grow at a normal rate, but shed at a slow rate.
- Shed faster than they grow.

There are different types of inheritance patterns of ichthyosis, including:

- Dominant, which means you inherit one normal copy and one mutated copy of the gene that causes ichthyosis. The abnormal copy of the gene is stronger or “dominant” over the normal copy of the gene, causing the disease. A person with a dominant mutation has a 50% chance (1 in 2) of passing the disorder to each of his or her children.
- Recessive, which means that your parents do not have signs of ichthyosis, but both parents carry only one abnormal gene, which is not enough to cause the disease. When both parents carry one recessive gene, there is a 25% chance (1 out of 4) per pregnancy of having a child who inherits both of these mutated genes and develops the disorder. There is a 50% chance (2 out of 4) per pregnancy of having a child who inherits only one mutated recessive gene, making them a carrier of the disease gene without noticeable signs. If one parent has a recessive form of ichthyosis with two mutated genes, all their children will carry one abnormal gene, but will not usually have noticeable signs of ichthyosis.

- X-linked, which means the gene mutations are located on the X sex chromosome. Each person has two sex chromosomes: Females generally have two X chromosomes (XX), and males generally have one X chromosome and one Y chromosome (XY). The mother always passes on an X chromosome, but the father can pass on either an X or Y chromosome. The inheritance pattern for X-linked ichthyosis is usually recessive; this means that males, who only have one X chromosome to begin with, pass on the mutated X chromosome. Because of this pattern, females are affected more often, and typically they have one mutated and one normal X chromosome.
- Spontaneous, which means the gene mutation occurs randomly without a family history of the disorder.

Diagnosis of Ichthyosis

Health care providers usually diagnose ichthyosis by:

- Asking about your family and medical history, including any skin disorders.
- Completing a physical exam, which includes a close examination of the skin, hair, and nails.
- Performing a skin biopsy to examine the tissue under a microscope. Sometimes doctors use a biopsy to help diagnose the condition or determine if the symptoms are from another disease or skin condition.

In addition, your health care provider may be able to diagnose ichthyosis with a genetic test that detects the mutated gene usually from a blood sample or a swab from the mouth. A genetic counselor or specialist can help you understand the test results.

Treatment of Ichthyosis

There is currently no cure for ichthyosis. The goals of treatment include reducing the redness of the skin, thickness of the scales, and itching. Treatments can include:

- Hydrating the skin with creams, lotions, or ointments to help trap moisture in the skin and relieve dryness and scaling. This works best if the topical agents are applied when the skin is moist.
- Taking long baths to soften and release scales.
- Taking a retinoid, a type of medication that can decrease scaling.
- Using prescription creams or ointments that may contain retinoids or other medications.

Depending on the type and severity of the disease, doctors may recommend additional treatment with “keratolytic” topical agents, which can help to loosen scales. However, these can be irritating for some people and have potential side effects if used in large amounts. Talk to your doctor before using any treatment option.

Who Treats Ichthyosis?

You may see one or more of the following specialists:

- Dermatologists, who specialize in conditions affecting the skin, hair, and nails.
- Clinical geneticists, who diagnose and treat children and adults with genetic disorders.
- Ophthalmologists, who treat disorders and diseases of the eye.
- Genetic counselors, who counsel and educate people on their genetic health.
- Audiologist, who diagnoses and treats hearing and balance problems.
- Nurse educators, who specialize in helping people to understand their overall condition and to set up their treatment plans.
- Pediatricians, who diagnose and treat children.
- Primary care physicians, who diagnose and treat adults.

Living With Ichthyosis

Depending on the type and severity of the disorder, you may find living with ichthyosis to be challenging. However, the following self-care tips may help you manage the disease, improve your health, and enjoy a better quality of life:

- Take baths to add more moisture to the skin and help remove the scales before applying topical agents.
- Keep your environment cool. This can help some people with ichthyosis who cannot tolerate heat, have reduced sweating, or have a lot of itching.
- Heat and air conditioning can produce very dry air. Using a humidifier can help keep moisture in the air and keep the skin from drying out as much.
- Wear loose-fitting clothes made from materials such as cotton, which may be less irritating to the skin.
- Use laundry detergents designed for sensitive skin that do not contain a lot of dyes or perfumes.
- Find a supportive community or join an online support group focused on ichthyosis. Some people may find it helpful to speak to a mental health professional about coping with the disorder.

Research Progress Related to Ichthyosis

The NIAMS supports basic science and translational and clinical research at universities and other organizations throughout the country. Researchers are studying ichthyosis to better understand the cause of the disease and to find better treatments, in hopes to better diagnose the disease, control its symptoms, and someday potentially cure the disorder.

Research topics include:

- Studying and identifying the genes linked to ichthyosis and their effect on the skin.
- Researching new treatments for the disorder, including topical, oral, or injected medications, or potentially designing newer genetic therapies.
- Looking at the normal development of the skin for clues as to how tissue development goes wrong to cause diseases like ichthyosis.

In 1994, the NIAMS funded the National Registry for Ichthyosis and Related Disorders. This registry voluntarily identifies people with ichthyosis and other related disorders to collect information about their skin disorder and how it has affected them. Although the original registry is now closed to new enrollment, it continues to provide crucial information to health care providers and scientists.

For More Info

U.S. Food and Drug Administration
Toll free: 888-INFO-FDA (888-463-6332)
Website: https://www.fda.gov

Drugs@FDA at https://www.accessdata.fda.gov/scripts/cder/daf. Drugs@FDA is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics
Website: https://www.cdc.gov/nchs

American Academy of Dermatology
Website: https://www.aad.org

Foundation for Ichthyosis and Related Skin Types, Inc.
Website: https://www.firstskinfoundation.org

National Registry for Ichthyosis and Related Disorders
Website: https://depts.washington.edu/ichreg/ichthyosis.registry

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

- Asian Language Health Information
- Spanish Language Health Information